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Case Report

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Ocular manifestations of atypical hemolytic uremic syndrome: a case report

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ABSTRACT

Atypical hemolytic uremic syndrome (aHUS) is a rare but serious condition that is often associated with renal impairment and thrombotic microangiopathy. This case report presents the case of a 20-year-old female with a long-standing diagnosis of aHUS who developed hypertensive choroidopathy, a rare ocular manifestation. The patient presented with sudden-onset hypertension and visual disturbances, with a blood pressure of 270/120 mmHg. Ophthalmological examination revealed bilateral papilledema, Elschnig spots, and Siegrist streaks, indicative of hypertensive choroidopathy. Imaging studies, including fundus photography, fluorescein angiography, and optical coherence tomography (OCT), confirmed retinal findings and optic disc edema. This case emphasizes the critical need for early detection and regular ophthalmological monitoring in aHUS patients, as ocular changes, such as hypertensive choroidopathy, can be early indicators of systemic complications. Early intervention, including blood pressure management, is essential for preventing irreversible damage to vision. This report underscores the importance of an integrated multidisciplinary approach to managing aHUS, especially considering the systemic nature of the disease and its potential ocular implications.

Keywords: Hypertensive choroidopathy, papilledema, atypical hemolytic uremic syndrome, Siegrist streaks, Elschnig spots

INTRODUCTION

Hemolytic uremic syndrome (HUS) is a disorder characterized by a triad of hemolytic anemia, thrombocytopenia, and acute kidney injury, typically triggered by infections, particularly those caused by Shiga toxin-producing Escherichia coli. Atypical hemolytic uremic syndrome (aHUS) is an uncommon, genetically heterogeneous condition primarily caused by dysregulation of the alternative complement pathway. Unlike typical HUS, aHUS can result in multiorgan involvement, most commonly affecting the kidneys, leading to thrombotic microangiopathy and renal failure.[1]

While aHUS mainly affects the kidneys, ocular involvement in aHUS can present with significant retinal changes, and hypertensive choroidopathy is one of the less recognized manifestations. Ocular impairment is rare but if present, it can be a serious complication of aHUS. According to some case report studies, sudden onset symptoms easily lead to near or total loss of vision.[2-4] In spite of initiating the full treatment some patients still can have these symptoms and their visual deficits become permanent.[2,3,5] Hypertensive choroidopathy refers to retinal changes resulting from long-standing or poorly controlled hypertension that can lead to ischemic damage to the retinal and choroidal vasculature. These changes can manifest as Elschnig spots, Siegrist streaks, and papilledema, which are indicative of hypertensive retinopathy and reflect a systemic vascular compromise. While these ocular signs are well documented in systemic hypertension, their presence in aHUS is less frequently reported.

This case report presents a 20-year-old female diagnosed with aHUS at an early age who developed hypertensive choroidopathy as a result of long-standing hypertension associated with her underlying disease. The patient exhibited significant ocular findings, including bilateral papilledema, Elschnig spots, and Siegrist streaks, which were detected during routine ophthalmologic examination following an episode of intracranial hemorrhage. This case underscores the importance of early recognition of ocular manifestations in aHUS, as timely intervention can prevent irreversible visual impairment and aid in the management of systemic complications.

Given the systemic nature of aHUS and the potential for multiorgan damage, it is essential for clinicians to include ophthalmological evaluations as part of the multidisciplinary care approach. Regular monitoring of retinal health, along with the management of blood pressure and renal function, is critical in preserving both visual function and overall patient health.

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CASE

A 20-year-old female with a long-standing diagnosis of aHUS, first diagnosed at the age of 10 years, was referred to our hospital for further evaluation after presenting with an intracranial hemorrhage. Her medical history included recurrent episodes of hemolytic anemia, thrombocytopenia, and renal impairment, for which she had been under regular follow-up and treatment at an external center. Despite being on standard therapy for aHUS, including eculizumab, the patient had not received treatment for the past 2-3 years due of intermittent non-compliance and complications related to her underlying condition.

The patient presented to the nephrology unit following sudden onset of severe headache, syncope, and loss of consciousness. Blood pressure at the time of admission was found to be dangerously elevated at 270/120 mmHg, and a CT scan revealed an intraventricular hemorrhage, confirming the diagnosis of a hemorrhagic cerebrovascular accident (CVA). The patient had no prior history of hypertension or cardiovascular disease, which led to further investigation into the underlying cause of her condition.

During her hospitalization, the patient also reported bilateral visual disturbances and a gradual decrease in visual acuity, which prompted a referral to the ophthalmology department.

Upon presentation to the ophthalmology clinic, the visual acuity was 20/60 in the right eye (OD) and 20/50 in the left eye (OS).

Anterior Segment

The anterior segment examination result was unremarkable, with no signs of cataracts or anterior uveitis.

Fundus Findings

After pupillary dilation, a detailed fundus examination revealed significant retinal changes in both eyes.

Grade 2 papilledema and increased vascular tortuosity were observed in the OD. Elschnig spots and Siegrist streaks were also observed in the peripheral retina. The macula appeared intact, with no visible hemorrhage, exudate, or edema (Figure 1).

Figure 1. Colour fundus photograph of the right eye (OD)

Grade 3 papilledema was observed in the OS, with similar findings of Elschnig spots and Siegrist streaks in the peripheral retina. No significant macular edema or hemorrhage was observed (Figure 2).

Figure 2. Colour fundus photograph of the left eye (OS)

Further diagnostic imaging was performed to evaluate the extent of retinal changes.

Fundus Autofluorescence (FAF)

FAF showed hypo-autofluorescence in the regions of Elschnig spots, indicating ischemic damage, and hyper-autofluorescence surrounding the ischemic areas due to hyperpigmentation (Figure 3).

Figure 3. Fundus autofluorescence (FAF) image of the right and left eyes (OD and OS)

Fluorescein Angiography (FFA)

FFA revealed early patchy leakage and delayed choroidal filling at the sites of Elschnig spots. Siegrist streaks showed areas of hypofluorescence consistent with choroidal filling defects (Figure 4).

Optical Coherence Tomography (OCT)

OCT demonstrated elevation of the optical disc, consistent with papilledema. However, there was no evidence of retinal detachment or macular edema. The foveal contour remained preserved, which is a positive indicator of the patient's retained central vision (Figure 5).

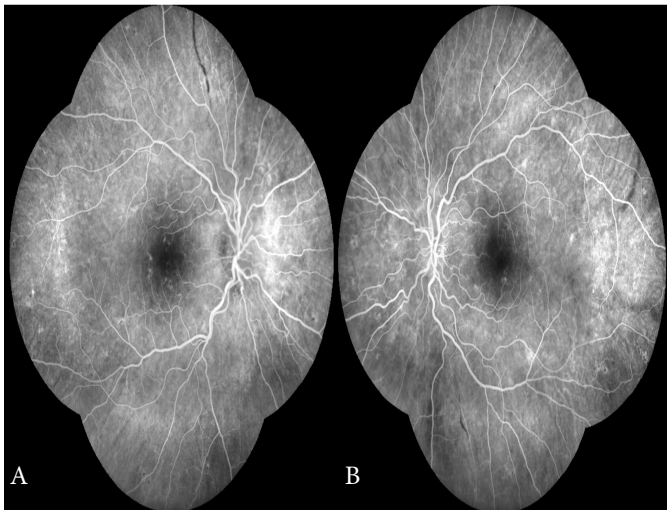

Figure 4. Fundus fluorescein angiography (FFA) of the right and left eyes (OD and OS)

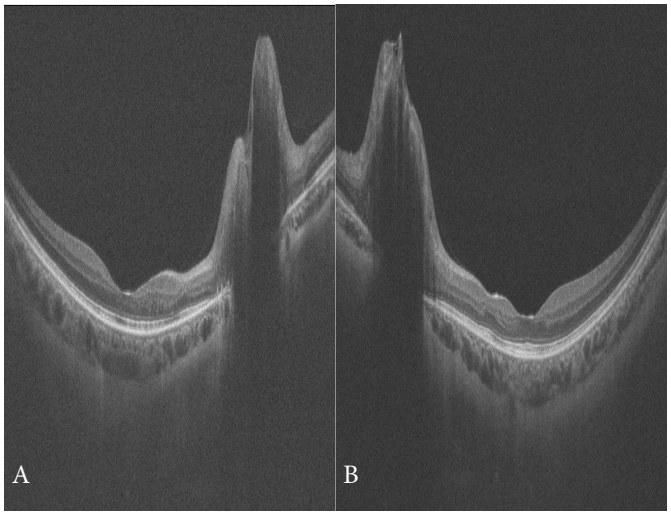

Figure 5. Optical coherence tomography (OCT) of the right and left eyes (OD and OS)

Upon confirming the diagnosis of hypertensive choroidopathy and papilledema associated with underlying aHUS, the patient was started on strict antihypertensive therapy to control her blood pressure, targeting a goal of <140/90 mmHg. Regular monitoring of renal function and continued treatment for aHUS were emphasized alongside scheduled follow-up visits with the ophthalmology department to track any changes in retinal findings.

The patient's condition was further managed in a multidisciplinary manner with nephrologists, ophthalmologists, and hematologists working together to optimize her care. Her blood pressure was gradually controlled, and follow-up fundus examinations showed no progression of retinal changes over the subsequent months. Regular ophthalmological examinations were scheduled to monitor the stability of her visual function and detect potential complications early.

At the time of her latest follow-up visit, the visual acuity remained stable at 20/60 in the OD and 20/50 in the OS. The papilledema had partially resolved, and the retinal findings of Elschnig spots and Siegrist streaks remained stable without any significant deterioration. Patients continue to be monitored for any systemic or ocular complications related to aHUS, with a strong emphasis on the importance of blood pressure control and regular retinal assessments.

DISCUSSION

aHUS is a rare genetically driven disorder primarily characterized by complement dysregulation, leading to systemic thrombotic microangiopathy. Although the renal manifestations of aHUS are well documented, the involvement of other organ systems, particularly the eyes, is less frequently reported. Ocular involvement in aHUS can range from subtle retinal changes to more severe conditions, such as hypertensive choroidopathy, which can lead to permanent vision loss if not diagnosed and managed early.

We present the case of a 20-year-old female with a longstanding diagnosis of aHUS who developed hypertensive choroidopathy in the context of poorly controlled hypertension. The patient exhibited Elschnig spots and Siegrist streaks, which are classic signs of hypertensive choroidopathy. These findings highlight the importance of regular ophthalmic evaluations in patients with aHUS, particularly those with long-term hypertension, as retinal findings can precede systemic complications and provide early indicators of disease progression.

Hypertensive choroidopathy, although commonly associated with systemic hypertension, is less frequently recognized in aHUS. Elschnig spots are small, round, pigmented areas of the retinal pigment epithelium that result from ischemia in the choriocapillaris, whereas Siegrist streaks are linear choroidal scars that reflect the chronicity of ischemic damage. In our case, both findings were observed in the periphery of the retina, indicating a significant vascular compromise. Papilledema, which was also noted in both eyes, is a direct result of elevated intracranial pressure, and can contribute to visual disturbances if left untreated.

FFA and FAF findings are crucial for confirming the ischemic nature of these retinal changes. The patchy leakage and delayed choroidal filling observed on FFA further supports the diagnosis of hypertensive choroidopathy. In addition, OCT allows for the visualization of papilledema without evidence of retinal detachment or macular edema, which is reassuring as it suggests the preservation of macular function and central vision.

Given the systemic nature of aHUS, this case highlights the need for an integrated approach to patient care involving not only nephrologists and hematologists, but also ophthalmologists. The early diagnosis of ocular changes can help prevent irreversible damage and preserve vision. Regular monitoring of blood pressure, renal function, and retinal health is essential in managing patients with aHUS, especially because they may not exhibit overt symptoms until significant complications arise.

Our case adds to the limited literature on the ocular manifestations of aHUS, particularly hypertensive choroidopathy. This underscores the importance of ophthalmic evaluation as part of the routine management of aHUS, especially in patients with long-term hypertension or those undergoing complement inhibitor therapy, as these patients are at an increased risk for vascular damage in multiple organ systems.

CONCLUSION

This case report illustrates the critical role of the early recognition and management of ocular manifestations in patients with aHUS. The presence of hypertensive choroidopathy in this patient, as evidenced by Elschnig spots and Siegrist streaks, highlights the need for routine ophthalmological assessment, particularly in patients with long-term hypertension. Timely intervention, including blood pressure control and regular monitoring of retinal health, is crucial to prevent irreversible visual impairment and manage the systemic complications of aHUS.

The findings of this case underscore the systemic nature of aHUS, emphasizing the importance of a multidisciplinary approach in managing these patients. Ophthalmologists, nephrologists, and hematologists must work together to ensure comprehensive care, allowing for the early detection of complications, improved patient outcomes, and preservation of both renal and visual functions.

ETHICAL DECLARATIONS

Informed Consent

The patient signed and free and informed consent form.

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Financial Disclosure

The authors declared that this study has received no financial support.

Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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